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Molecular diagnosis in patients with retinoblastoma: Report of a series of cases.

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Abstract

Objectives: To report the benefits of genetic diagnosis in patients with retinoblastoma.

Method: Observational study. Patients with retinoblastoma and their families were included. Demographic and clinical data were recorded. Blood and tumor samples were obtained. Next generation sequencing was performed on the samples. When deletion 13 q syndrome was suspected, cytogenetics microarray was performed (Cytoscan® HD, Affymetrix, Santa Clara, CA, USA), with a high density chip of 1.9 million of non-polymorphic probes and 750 thousand SNP probes.

Results: Of the 7 cases were analyzed 4 were male. The mean age at diagnosis was 21 months (range 5‑36). Three cases had bilateral retinoblastoma, and 4 unilateral. None had family history. In all patients, blood was analyzed, and a study was performed on the tissue from 2 unilateral enucleated tumors, in which 6 mutations were identified, all *de novo.* Just one was novel (c.164delC; case 1). One case of unilateral tumor revealed blood mosaicism, showing that his condition was inheritable, and that there is a high risk of developing retinoblastoma in the unaffected eye. The patient also has an increased risk of presenting with other primary tumors.

Conclusion: Molecular diagnosis of *RB1* in patients with retinoblastoma impacts on the decision process, costs, treatment, and prognosis of patients, as well as their families.